Genomics: How we can all play a role in shaping the future of health research

Jack Nunn

In the near future the affordability of genomic sequencing will make it as routine as a blood test, offering people a new frontier in understanding their own identity in relation to genomics and treatment pathways.

Around the world, genomics developments of profound potential are already occurring. These range from extending individuals' knowledge of their own genetic features, to enabling them to have their genetic makeup available for research, to contacting others who may share certain genes or mutations.

However, without a clear ethical framework to guide the prioritisation of research and treatments, there is a risk that profitability will be the stronger force in influencing the shape of the research agenda, and thus, treatments to diseases which are available to us all.

We need to take a giant imaginative leap to ensure that the public remains involved in every stage of genomics research, invited to be active partners in shaping the future of research that can improve lives.

Australia urgently needs to work with the public and across international borders to support innovative ways to harness the positive potential of the genomic era, while ensuring that the research remains prioritised on the needs of people.

Involving the public in research

The National Health and Medical Research Council defines public involvement as:

"research being carried out with or by consumers and community members rather than to, about or for them".

To involve the public in research means that people can influence each stage of research - from identifying what should be researched, prioritising which of these topics should be funded right through to doing the research or helping disseminate and translate the findings.

I will avoid the word 'consumer'. I will use 'the public' or 'people', as I feel this implies an equal partnership. The Health Research Authority in England came up with what I think is the best description of 'the public':

"When we use this term, public means patients, potential patients or members of the public including those with known genetic dispositions, carers and people who use health and social care services as well as people from organisations that represent people who use health and social care services."[1]

Involving the public in science, specifically genomic research, allows us to walk the line between two poles which UNESCO describes as 'morbid distrust and blind confidence'[2].

Similarly there is often a 'disconnect between the views of those handling the findings of research and those participating in research', and involving the public in how the research is done is an essential stage in working through this disconnection in an ethical way.

Re-imagining public involvement

Just as there has been a shift in bio-banking from seeing donors as participants to seeing the public as partners and 'collaborators', the same shift must happen with genomic data repositories.

"The role of patients and the public in bio-banking activities has been viewed traditionally as bio-bank participants rather than as collaborators in the design, development and ongoing operation and governance of bio-banks. However, there is growing acceptance and appreciation of the value that patients, patient advocacy organisations and the public can bring as stakeholders in bio-banking and more generally in research."[3]

In genomics, this is a mental shift comparable to books being digitised - with the physical books still precious, but the fundamental data shareable and open to analysis by anyone in ways that have never before been possible.

The Wiki-genome

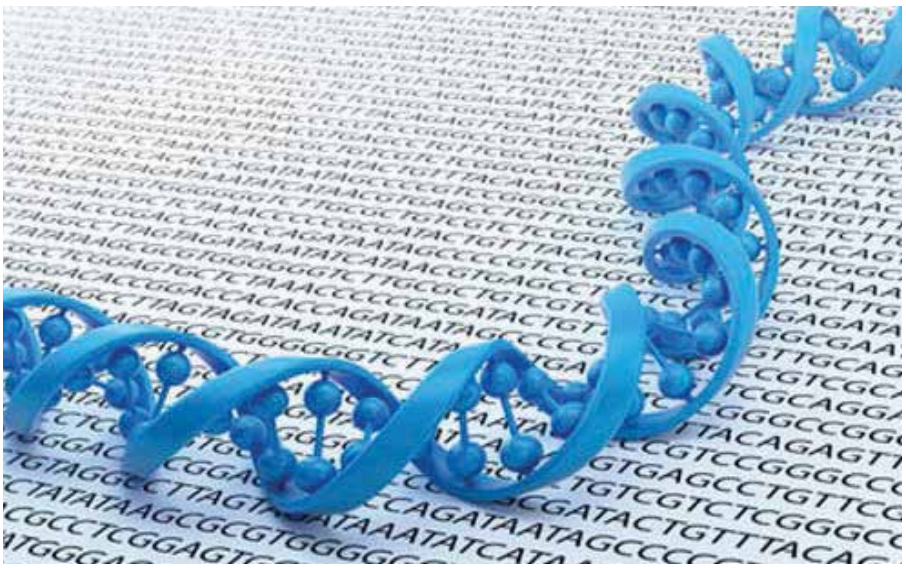
Social media is a powerful tool. Let's agree that broadly speaking, Wikipedia is a social media tool. What if we took the same approach to human knowledge and applied it to genomics?

Let's imagine a hypothetical scenario. The Wikimedia Foundation, or another not-for-profit organisation with good intentions, sets up a free online service where anyone can upload their entire genome (when compressed it is around 4mb, this is less than a high-resolution photo or about the size of song download). Let's assume they have unbreakable security and encryption. The service provides free learning resources in every known human language to support people to make informed decisions.

Please note that everything described below is happening somewhere already, just not necessary as part of the same linked-service.

The free hypothetical service offers a number of options and choices, which may be altered at any point:

- **Full genome analysis:** they will interpret your genome with the most up-to-date analysis available - looking for 'known significances'. You can choose what level of information you would like to know, ranging from 'nothing' to 'everything'.
- **Full genome sharing:** You are invited to submit your genome so that it can be accessed by researchers who have gained ethical approval. You may even chose sub-sets of research you are happy to share your genome for if it is not a case of 'select all'.
- **Phenotype, lifestyle and medical history sharing:** There is an option for people to complete surveys or



pre-approved data fields that would be of use and interest to researchers now and in the future. Many aspects of this would involve self-identifying.

- **Contact:**
  - You are offered the choice to find other people on the service who may share certain genes or mutations with you, or others including researchers to contact you. This may include the option of finding family members (much as many for-profit ancestry websites currently do). This may involve invitations to be participants in research.
- **Involvement:** You are invited to get involved in shaping the future of research.
  - You can search for and join or start discussions with the public and researchers about research which is planned or already taking place.
  - You may work with researchers others to identify and prioritise areas of research and work with others to help generate and evaluate outcomes.
- **Researchers** with approved credentials could, if they wished, be contacted by those in the service about their research and be alerted when people with certain genes or mutations joined the service, and recruit participants to research.

This hypothetical free service may never materialise, but it raises important ethical questions that need answering. Crucially, it also gives a real and achievable mechanism for people to get involved in genomic research in a way which is systematic, open access, free and universal.

This scenario doesn't answer who will pay for the sequencing itself, but it is likely that the cost of having a 'direct to consumer' test will become significantly more affordable to a greater number of people in the near future. Similarly, full genome sequencings will be offered by some health services for free in Australia in the near future, as routinely as a blood test is now.

Next steps

There are a number of organisations trying to improve genomic research and involve the public (DNADigest, GenomesConnect), but the more people who are aware of how they can get involved, the greater the chance of future research meeting the needs of the public.

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1 http://web.archive.org/web/20160309063134/http://www.hra.nhs.uk/documents/2013/10/hra-public-involvement-strategy-circulation-september-2013.pdf
2 http://web.archive.org/web/20160309063045/https://depot.erudit.org/bitstream/003001dd/1/Involving_the_Public_in_Public_Health_Genomics.pdf
3 http://link.springer.com/article/10.1186/s40900-015-0001-z/fulltext.html